



COL5A1 gene

collagen type V alpha 1 chain

Normal Function

The *COL5A1* gene provides instructions for making a component of type V collagen. Collagens are a family of proteins that strengthen and support many tissues in the body, including skin, ligaments, bones, tendons, and muscles.

A component of type V collagen called the pro- α 1(V) chain is produced from the *COL5A1* gene. Collagens begin as rope-like procollagen molecules that are each made up of three chains. Two combinations of chains can produce type V collagen: three pro- α 1(V) chains or two pro- α 1(V) chains and one pro- α 2(V) chain (which is produced from the *COL5A2* gene).

The triple-stranded procollagen molecules are processed by enzymes outside the cell to create mature collagen. The collagen molecules then arrange themselves into long, thin fibrils with another form of collagen, type I. Type V collagen regulates the width (diameter) of those fibrils. Studies suggest that type V collagen also controls the assembly of other types of collagen into fibrils in several tissues.

Health Conditions Related to Genetic Changes

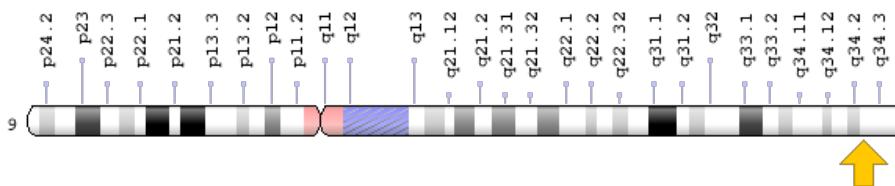
Ehlers-Danlos syndrome

Mutations in the *COL5A1* gene cause a form of Ehlers-Danlos syndrome called the classical type. Ehlers-Danlos syndrome is a group of disorders that affect the connective tissues that support the skin, bones, blood vessels, and many other organs and tissues. This form of the disorder is characterized by skin that is soft, highly stretchy (elastic), and fragile; abnormal scarring; and an unusually large range of joint movement (hypermobility). More than 100 *COL5A1* gene mutations have been identified in people with classical Ehlers-Danlos syndrome. The mutations affect one copy of the gene in each cell, reducing the amount of pro- α 1(V) chains that cells produce. As a result, fibrils containing type V and type I collagens in the skin and other tissues are disorganized and larger than usual. Researchers believe that the abnormal collagen weakens connective tissues throughout the body, which causes the signs and symptoms of classical Ehlers-Danlos syndrome.

Chromosomal Location

Cytogenetic Location: 9q34.3, which is the long (q) arm of chromosome 9 at position 34.3

Molecular Location: base pairs 134,641,790 to 134,844,843 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- alpha 1 type V collagen preproprotein
- CO5A1_HUMAN
- collagen type V alpha 1
- collagen, type V, alpha 1

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Collagen: The Fibrous Proteins of the Matrix
<https://www.ncbi.nlm.nih.gov/books/NBK21582/>

GeneReviews

- Ehlers-Danlos Syndrome, Classic Type
<https://www.ncbi.nlm.nih.gov/books/NBK1244>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28COL5A1%5BTIAB%5D%29+OR+%28collagen%5BTI%5D%29+AND+%28type+V%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

OMIM

- COLLAGEN, TYPE V, ALPHA-1
<http://omim.org/entry/120215>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_COL5A1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=COL5A1%5Bgene%5D>
- Ehlers-Danlos Syndrome Variant Database
https://eds.gene.le.ac.uk/home.php?select_db=COL5A1
- HGNC Gene Family: Collagens
<http://www.genenames.org/cgi-bin/genefamilies/set/490>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2209
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1289>
- UniProt
<http://www.uniprot.org/uniprot/P20908>

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